


Informed Consent for Genetic Testing


**Patient Education information
for use with the Michigan Model
Consent Form for Genetic
Testing**



Many new advances are occurring in medicine, especially in the field of medical genetics. This booklet was prepared by the Michigan Department of Community Health to help answer your questions about genetic testing. It tries to make sure you are well informed before you agree to give a sample for testing. A glossary is included at the back to help you understand genetics-related terms.

Genetic testing is voluntary. Only you can decide if you want to be tested. Most genetic conditions are complex and each person's situation is unique. You may want more information before you decide what to do. People often have questions about the condition itself or other issues such as insurance coverage. Your health care provider will explain the purpose of the test and try to answer your questions.

If you still have unanswered questions, be sure to ask your physician, nurse, or genetic counselor before you agree to have the genetic test being offered to you.



1. What is a genetic test?



There are many different types of genetic tests. A genetic test is any analysis used to look at a person's genetic make-up. The genes a person is born with may cause a disease or birth defect. They can also cause certain physical features, or an increased chance of developing a certain condition. Genetic tests are usually performed on samples of blood, tissue or cheek cells. When testing a fetus, chorionic villus cells or amniotic fluid can be used. The test may examine a person's deoxyribonucleic acid (DNA), ribonucleic acid (RNA), proteins, or other chemicals in cells that can indicate a genetic condition. The type of test performed depends on the reason for testing. Genetic tests can be used to confirm a diagnosis. They can help predict the chance a person will develop a disease or condition in the future. They can also be used as a method of carrier screening to find out if a person has specific genes that increase the chance of a disease or birth defect occurring in their children.

2. What are the limitations of genetic testing?

Current genetic tests cannot provide a satisfactory answer for everyone concerned about a genetic disease or birth defect. There is no one test that detects all genetic diseases. Most genetic tests are specific only for the condition discussed with you and listed on your consent form. Genetic testing is highly accurate, but there are limitations. For example:

▪ Errors

Information provided by you, your physician, or medical records may not be totally accurate. It is very important to have correct information. If a person is being tested to rule out the same disease that a relative has, the laboratory needs to know *exactly* which test to perform. If your relative really had a different disease than the one you are tested for, your own result will not give accurate information about whether you have the condition. Tests that depend on having samples from relatives may be interpreted incorrectly if the true bloodlines in a family are not known.

- **Laboratory processing**

In rare cases, it is possible for something to go wrong in handling the sample. This might lead to incorrect results. Examples include sample mislabeling, sample contamination, or misinterpretation of laboratory findings. Sometimes the laboratory may need a second sample in order to finish the test.

- **Implications of results**

The test may reveal a genetic abnormality, but cannot always predict with certainty whether a person will develop a disease, how severe the condition will be, or when symptoms will appear.

3. What are the benefits and risks of genetic testing?

Benefits: There are benefits to genetic testing. A person found to have an increased risk of disease might want to choose preventive or therapeutic medical treatments. Having the knowledge can empower a person and family members to make important life planning decisions, even if a cure is not available at that time. Having a specific diagnosis could qualify a person to enroll in research studies, which may lead to new treatments. Knowing about a certain disease gene might provide important health information for a person's family.



Risks: The physical risk of testing is usually minimal, typically not more than providing a blood sample. If your test involves any other type of sample, the physician performing the procedure, or a designated representative, should explain the risks before you decide to have the test.

The greatest concern pertains to the way a genetic test result might change a person's life. The decision to have genetic testing can be very stressful. You may have strong emotional reactions to learning that you do - or do not - carry the gene for a certain condition. For these reasons, a psychological evaluation may be indicated before

some genetic tests are performed. Sometimes a positive test result can affect family relationships. A person who decides to have genetic testing needs to consider whether to inform other family members. Sometimes the result for one family member can disclose information about the genetic make-up of other relatives, even if they have not been tested. Furthermore, a genetic test may reveal unexpected relationships, such as non-paternity (a different biological father).

A genetic test result may also affect a person's ability to obtain health, life, and disability insurance. It could also affect the ability to obtain or keep a job. In Michigan, Public Acts 26, 27, 28, and 32 of 2000 help to protect citizens from discrimination on the basis of a genetic test result. The laws make it illegal for companies to require genetic testing as a condition of employment or health insurance. Life insurance and disability insurance are not included in the Michigan laws. If a genetic test is performed, your insurance company may have access to the result.

4. What do the results of genetic testing mean?

Genetic tests are often a valuable tool, but sometimes do not provide a definite answer. Most often, results can be classified as negative or positive.



Negative or “normal”: Usually a negative test is good news, but there are some cautions. Some clinical conditions can be caused by many different

forms of a gene. This means a negative test does not always rule out the presence of every gene capable of causing that disease. You still might carry one of these other genes, even if your test is normal. Most laboratories only test for the most common forms. It is possible that a person could carry one of the rare genes not included in the test. If you receive a negative result now, it may still be important to check with your physician or genetics clinic to determine if better tests are available in the future.

Positive or “abnormal”: A positive or “abnormal” result means you carry a form of gene that can cause a specific condition, or can put you at increased risk of developing a disease. However, it may or may not mean you have, or will get the disease. Depending on the disorder, and gene form, there may not be any effects.

Inconclusive: Sometimes a test result falls in between the normal and abnormal range. In such cases, it may not be possible to tell whether a person carries a gene that can cause a disease.

Be sure you understand what a negative or positive test result will mean by discussing your situation with your physician, nurse, or genetic counselor.

5. What will happen to my sample after the genetic test is completed?

It depends on the laboratory that performs your test. Some laboratories have their own consent form (in addition to this one) that allows you to say what you want done with the sample. Many laboratories keep leftover samples. These may be used for scientific purposes or medical research, or as “controls” to insure the accuracy of laboratory methods. Laboratories may re-run a test when new technology becomes available. Unless you are specifically submitting a sample for DNA banking, there is no guarantee it would be available to you or your family for future testing.

6. Who has access to my sample and information from the test?

Genetic tests are handled in a confidential manner, like other personal health information. The person collecting your sample, and people in the laboratory performing the test, will need to handle your sample. Your medical record and test results are confidential. Your provider may ask for your written permission before releasing any results to third parties.

7. What is the cost of genetic testing?



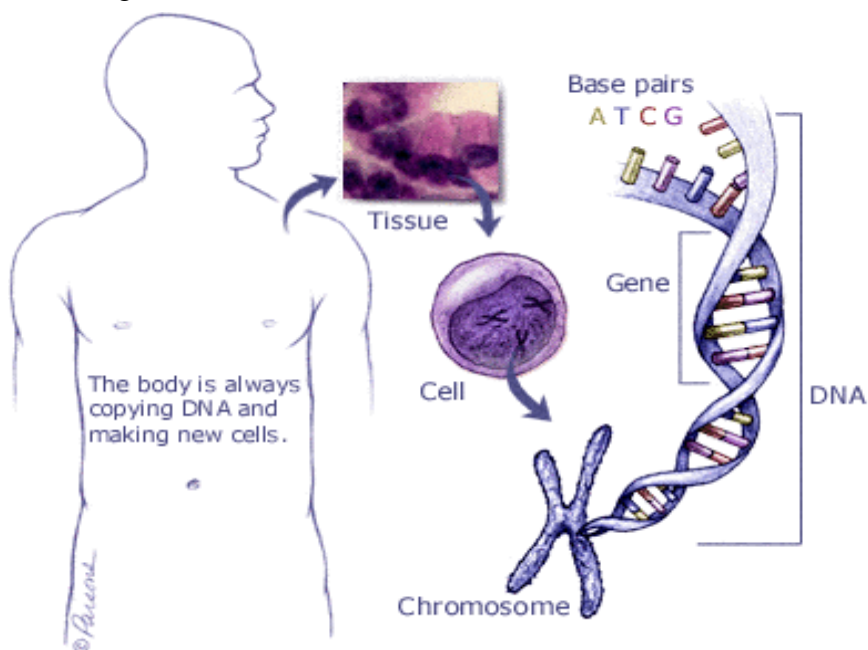
The cost depends on the test performed and number of family members tested. Typically the cost can range from a few hundred to a few thousand dollars. Before being tested, ask about the cost of the test and whether it is covered by insurance. Find out if you will be responsible for some or all of the bill. Genetic tests are not always covered by medical insurance, and you may need to pay some or all of the cost.

GLOSSARY OF GENETIC TERMS

Amniotic fluid: The fluid surrounding an unborn fetus; it contains fetal cells that can be used for genetic testing.

Birth defect: An abnormal condition that occurs before or at birth. Birth defects can cause physical and developmental problems that may require special medical care or therapy.

Carrier status: The knowledge of whether a healthy person carries a disease-causing gene that could be passed down to his or her children. Carrier status is often determined by genetic screening.



Source: <http://pathology2.jhu.edu/pancreas/causespc.cfm>

Cell: A small membrane-bound compartment filled with chemicals; the subunit of all living things, including humans.

Chorionic villus cells: Cells contained in the tissue of the placenta in early pregnancy. The tissue contains the same genetic information as the developing fetus.

Chromosomes: Structures within the cell that store and transmit genetic information. A normal human cell contains 22 pairs of non-sex chromosomes, and two sex chromosomes.

Diagnostic testing: A test to confirm the presence or absence of a specific disease or condition.

DNA: Deoxyribonucleic acid - A large molecule contained in the chromosomes that carries all of the genetic information needed to operate a cell, make tissues and control organ systems.

DNA banking: The process of preserving and saving a person's DNA sample for future testing.

Enzyme: A protein that speeds up a biochemical reaction. Enzymes are critical to cell functions.

Fetus: An unborn baby from about eight weeks after conception until birth.

Gene: A subunit of DNA that contains the message for a cell product, typically some type of protein such as an enzyme. Humans have approximately 25,000 genes.

Genetic: A trait or condition determined by one or more genes.

Genetic counseling: The process of giving a person information about a genetic disorder or birth defect. Genetic counseling usually includes helping a person understand the medical facts, how heredity is involved, the risk to family members, and what options are available to deal with the risk, including available treatments. It also helps a person decide what course of action to take in view of his or her genetic risk, family goals, ethical and religious beliefs.

Informed consent: A person's agreement to allow a medical test, treatment or procedure based on a full understanding of all the facts necessary to weigh the benefits and risks of the proposed test, and make an intelligent decision.

Molecule: A chemical combination of two or more atoms that form a specific chemical substance; the smallest unit of a substance that displays characteristic physical and chemical properties.

Predictive testing: A genetic test to determine if a person has one or more genes that increase the risk of developing a certain disease or disability at some time in the future.

Prenatal testing: A genetic test performed on a fetus.

Pre-symptomatic testing: A genetic test performed before the onset of any symptoms to determine if a person has a gene that will eventually cause a certain disease or disability.

Protein: A large complex molecule essential to body structure, function and regulation. Examples include hormones, enzymes, and antibodies. Proteins are determined by the DNA sequences within genes.

RNA: Ribonucleic Acid - A cell molecule similar to DNA. It plays an important role in making proteins and other cell activities.

Screening: The process of looking for a particular gene or disease in individuals who don't exhibit any signs or symptoms.

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CONSENT TO OBTAIN A SPECIMEN FOR GENETIC TESTING

PATIENT LAST NAME: _____ (Please Print)		FIRST NAME: _____	MI: _____
DATE OF BIRTH: / /		HOSPITAL/ ID NUMBER: _____	
ORDERED BY: _____		I REQUEST GENETIC TESTING FOR: _____	
LABORATORY NAME AND ADDRESS: _____ _____ _____ _____		(name of condition)	
THERE IS A FAMILY HISTORY OF THIS CONDITION: q Yes q No		The intended purpose is (check all that apply):	
<div style="text-align: center;">SAMPLE TYPE</div> <div style="display: flex; flex-wrap: wrap;"><div style="width: 50%;"><input type="checkbox"/> Amniotic fluid <input type="checkbox"/> Blood <input type="checkbox"/> Cheek swab <input type="checkbox"/> Chorionic villus sample (CVS) <input type="checkbox"/> Skin <input type="checkbox"/> Tissue block <input type="checkbox"/> Other _____</div><div style="width: 50%;"><input type="checkbox"/> Carrier status <input type="checkbox"/> Diagnostic <input type="checkbox"/> Predictive <input type="checkbox"/> Prenatal <input type="checkbox"/> Pre-symptomatic <input type="checkbox"/> Screening <input type="checkbox"/> Other _____</div></div>			
<ol style="list-style-type: none">1. I have been informed about the purpose of this genetic test.2. I have received an explanation of the limitations of this genetic test.3. I have discussed the benefits and risks of this genetic test with my physician and other health care professional. I understand some genetic tests can involve possible medical, psychological and insurance issues for my family and me.4. I understand the meaning of possible test results and have been informed how I will receive the result.5. I understand any leftover sample may be retained by the laboratory, but only used for my future clinical care, with my permission. I understand I may cancel this consent up until the time the test is performed.6. I have been informed who may have access to the biological sample and genetic test result. I have been informed that the test result will be part of my medical record and remain confidential in accordance with standard medical practice.7. I have read this consent form and booklet. I received a copy of the form and booklet for my records. My questions have been answered to my satisfaction.			
<p>By my signature below, I give consent to have a sample taken for genetic testing on the above-named patient for the condition(s) listed above.</p> <div style="text-align: center; margin-top: 20px;">_____ <i>Signature of Patient or Authorized Designee</i></div> <div style="text-align: right; margin-top: 5px;">_____ Date</div> <div style="margin-top: 10px;"><div style="display: flex; justify-content: space-between;">Circle one:SelfParent(s)Legal GuardianDurable Power of Attorney for Health Care</div></div>			
Print Name of Authorized Person Reviewing Consent Form with Patient: _____			
Signature of Authorized Person: _____		Date: _____	

This consent form was developed by the Michigan Department of Community Health in compliance with PA 29 of 2000. It may not be altered or deleted to change the meaning of specific statements above or the intent of the informed consent process.